

Diffuse midline glioma H3 K27M-mutant diagnosis

Microscopic appearance

H3 K27M-mutant diffuse midline gliomas usually appear histologically as [astrocytic tumors](#). In a minority of cases they appear histologically low-grade (without mitotic figures, microvascular proliferation, or necrosis), however, even then they are considered WHO grade IV tumors.

Immunophenotype

[S100](#): positive

[NCAM1](#): positive

[OLIG2](#): positive

H3F3A K27M mutation: usually positive, although other mutations are also recognized

[p53](#) protein: positive in 50%

[GFAP](#): variable

chromogranin-A: negative

[NeuN](#): negative

Genetics

Genomic work has uncovered distinct mutations found in the majority of diffuse midline gliomas resulting in the inclusion of diffuse midline glioma H3 K27M-mutant as a distinct entity and the removal of diffuse intrinsic pontine gliomas from the current WHO classification of CNS tumors.

These mutations are in the histone H3F3A gene (K27M mutations) or less frequently HIST1H3B and HIST2H3C genes.

A number of other distinct but related histone gene mutations have also been identified in similar tumors.

Radiographic features

MRI

Diffuse midline glioma H3 K27M-mutant MRI.

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